



Modeling human disease in humans: the ciliopathies.

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Public Summary:

Soon, the genetic basis of most human Mendelian diseases will be solved. The next challenge will be to leverage this information to uncover basic mechanisms of disease and develop new therapies. To understand how this transformation is already beginning to unfold, we focus on the ciliopathies, a class of multi-organ diseases caused by disruption of the primary cilium. Through a convergence of data involving mutant gene discovery, proteomics, and cell biology, more than a dozen phenotypically distinguishable conditions are now united as ciliopathies. Sitting at the interface between simple and complex genetic conditions, these diseases provide clues to the future direction of human genetics.

Scientific Abstract:

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